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Noonan Syndrome With Multiple Lentiginos: Subtle Key Skin Clues to the Diagnosis[☆]



Síndrome de Noonan con lentiginosis: manifestaciones dermatológicas sutiles y claves

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A 15-year-old woman was evaluated for generalized redness and roughness of the skin that had been present since childhood. She had congenital pulmonary valve stenosis, incomplete pyeloureteral duplication of the right kidney, and learning difficulties. Physical examination revealed multiple lentiginos on the face and neck, about 100 melanocytic nevi, 4 nevus spilus-like spots, a depressed nasal bridge, and a broad philtrum (Fig. 1). Other notable findings included short stature; sparse eyebrows; follicular hyperkeratosis of the face, arms, and thighs; curly hair; and phototype IV. Family members did not present any diseases of interest and shared none of the aforementioned findings. A genetic study

found that the patient was heterozygous for the c.1403 C>T (p.Thr486Met) mutation in PTPN11, which is associated with Noonan syndrome with multiple lentiginos (formerly known as LEOPARD syndrome).

Bessis et al¹ prospectively collected data on skin alterations in 34 patients genetically diagnosed with Noonan syndrome with multiple lentiginos. In addition to those described in our patient, the following signs can be observed: café noir spots, alopecia, temporary alopecia, ulerythema ophryogenes, palmoplantar hyperkeratosis, skin hypermobility, redundant skin in acral regions, ecchymosis, hyperhidrosis, and lymphedema.

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Figure 1 Clinical image showing multiple lentigines, keratosis pilaris, sparse eyebrows, a broad nasal philtrum, bulbous nose, and curly hair.

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Conflicts of interest

The authors declare that they have no conflicts of interest.

Reference

1. Bessis D, Miquel J, Bourrat E, Chiaverini C, Morice-Picard F, Abadie C, et al. Dermatological manifestations in Noonan syndrome: a prospective multicentric study of 129 patients positive for mutation. *Br J Dermatol.* 2019;180:1438–48, <http://dx.doi.org/10.1111/bjd.17404>.